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PUBMED 2594783
REMARK SEQUENCE FROM N.A., AND VARIANT CAIS MET-866.
REFERENCE 3 (residues 1 to 919)
AUTHORS Govindan,M.V.
TITLE Specific region in hormone binding domain is essential for hormone binding and trans-activation by human androgen receptor
JOURNAL Mol. Endocrinol. 4 (3), 417-427 (1990)
MEDLINE 90258935
PUBMED 2342476
REMARK SEQUENCE FROM N.A.
REFERENCE 4 (residues 1 to 919)
AUTHORS Chang,C.S., Kokontis,J. and Liao,S.T.
TITLE Structural analysis of complementary DNA and amino acid sequences of human and rat androgen receptors
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 85 (19), 7211-7215 (1988)
MEDLINE 89017168
PUBMED 3174628
REMARK SEQUENCE FROM N.A.
TISSUE=Prostate
REFERENCE 5 (residues 1 to 919)
AUTHORS Tilley,W.D., Marcelli,M., Wilson,J.D. and McPhaul,M.J.
TITLE Characterization and expression of a cDNA encoding the human androgen receptor
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 86 (1), 327-331 (1989)
MEDLINE 89098909
PUBMED 2911578
REMARK SEQUENCE FROM N.A.
TISSUE=Prostate
REFERENCE 6 (residues 1 to 919)
AUTHORS Marcelli,M., Tilley,W.D., Wilson,C.M., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.
TITLE Definition of the human androgen receptor gene structure permits the identification of mutations that cause androgen resistance: premature termination of the receptor protein at amino acid residue 588 causes complete androgen resistance
JOURNAL Mol. Endocrinol. 4 (8), 1105-1116 (1990)
MEDLINE 91155943
PUBMED 2293020
REMARK SEQUENCE FROM N.A.
TISSUE=Prostate
REFERENCE 7 (residues 1 to 919)
AUTHORS Chang,C.S., Kokontis,J. and Liao,S.T.
TITLE Molecular cloning of human and rat complementary DNA encoding androgen receptors
JOURNAL Science 240 (4850), 324-326 (1988)
MEDLINE 88178111
PUBMED 3353726
REMARK SEQUENCE OF 189-919 FROM N.A.
REFERENCE 8 (residues 1 to 919)
AUTHORS Trapman,J., Klaassen,P., Kuiper,G.G., van der Korput,J.A., Faber,P.W., van Rooij,H.C., Geurts van Kessel,A., Voorhorst,M.M., Mulder,E. and Brinkmann,A.O.
TITLE Cloning, structure and expression of a cDNA encoding the human androgen receptor
JOURNAL Biochem. Biophys. Res. Commun. 153 (1), 241-248 (1988)
MEDLINE 88240407
PUBMED 3377788
REMARK SEQUENCE OF 468-919 FROM N.A.
REFERENCE 9 (residues 1 to 919)
AUTHORS Hsiao,P.W., Lin,D.L., Nakao,R. and Chang,C.

TITLE The linkage of Kennedy's neuron disease to ARA24, the first identified androgen receptor polyglutamine region-associated coactivator

JOURNAL J. Biol. Chem. 274 (29), 20229-20234 (1999)

MEDLINE 99329028

PUBMED 10400640

REMARK INTERACTION WITH RAN.

REFERENCE 10 (residues 1 to 919)

AUTHORS Sleddens,H.F.B.M., Oostra,B.A., Brinkmann,A.O. and Trapman,J.

TITLE Trinucleotide repeat polymorphism in the androgen receptor gene (AR)

JOURNAL Nucleic Acids Res. 20, 1427-1427 (1992)

REMARK POLYMORPHISM OF POLY-GLN REGION.

REFERENCE 11 (residues 1 to 919)

AUTHORS Lu,J. and Danielsen,M.

TITLE Direct Submission

JOURNAL Submitted (~FEB-1995)

REMARK POLYMORPHISM OF POLY-GLY REGION.

TISSUE=Blood

REFERENCE 12 (residues 1 to 919)

AUTHORS Giovannucci,E., Stampfer,M.J., Krithivas,K., Brown,M., Dahl,D., Brufsky,A., Talcott,J., Hennekens,C.H. and Kantoff,P.W.

TITLE The CAG repeat within the androgen receptor gene and its relationship to prostate cancer

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 94 (7), 3320-3323 (1997)

MEDLINE 97250535

PUBMED 9096391

REMARK POLYMORPHISM OF POLY-GLN REGION.

REFERENCE 13 (residues 1 to 919)

AUTHORS Giovannucci,E., Stampfer,M.J., Krithivas,K., Brown,M., Dahl,D., Brufsky,A., Talcott,J., Hennekens,C.H. and Kantoff,P.W.

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 94, 8272-8272 (1997)

REMARK ERRATUM.

REFERENCE 14 (residues 1 to 919)

AUTHORS Pinsky,L., Trifiro,M.A., Kaufman,M., Beitel,L.K., Mhatre,A., Kazemi-Esfarjani,P., Sabbaghian,N., Lumbroso,R., Alvarado,C., Vasilidou,M. and Gottlieb,B.

TITLE Androgen resistance due to mutation of the androgen receptor

JOURNAL Clin Invest Med 15 (5), 456-472 (1992)

MEDLINE 93092459

PUBMED 1458719

REMARK REVIEW ON VARIANTS.

REFERENCE 15 (residues 1 to 919)

AUTHORS Brown,T.R., Scherer,P.A., Chang,Y.-T., Migeon,C.J., Ghirri,P., Murono,K. and Zhou,Z.

TITLE Molecular genetics of human androgen insensitivity

JOURNAL Eur. J. Pediatr. 152 Suppl. 2, S62-S69 (1993)

REMARK REVIEW ON VARIANTS AIS.

REFERENCE 16 (residues 1 to 919)

AUTHORS Sultan,C., Lumbroso,S., Poujol,N., Belon,C., Boudon,C. and Lobaccaro,J.M.

TITLE Mutations of androgen receptor gene in androgen insensitivity syndromes

JOURNAL J. Steroid Biochem. Mol. Biol. 46 (5), 519-530 (1993)

MEDLINE 94059770

PUBMED 8240973

REMARK REVIEW ON VARIANTS.

REFERENCE 17 (residues 1 to 919)

AUTHORS Patterson,M.N., Hughes,I.A., Gottlieb,B. and Pinsky,L.

TITLE The androgen receptor gene mutations database

JOURNAL Nucleic Acids Res. 22 (17), 3560-3562 (1994)
MEDLINE 95023089
PUBMED 7937057
REMARK REVIEW ON VARIANTS.
REFERENCE 18 (residues 1 to 919)
AUTHORS Brinkmann,A.O., Jenster,G., Ris-Stalpers,C., van der Korput,J.A.,
Bruggenwirth,H.T., Boehmer,A.L. and Trapman,J.
TITLE Androgen receptor mutations
JOURNAL J. Steroid Biochem. Mol. Biol. 53 (1-6), 443-448 (1995)
MEDLINE 95352489
PUBMED 7626493
REMARK REVIEW ON VARIANTS.
REFERENCE 19 (residues 1 to 919)
AUTHORS Gottlieb,B., Trifiro,M., Lumbroso,R. and Pinsky,L.
TITLE The androgen receptor gene mutations database
JOURNAL Nucleic Acids Res. 25 (1), 158-162 (1997)
MEDLINE 97169385
PUBMED 9016528
REMARK REVIEW ON VARIANTS.
REFERENCE 20 (residues 1 to 919)
AUTHORS Veldscholte,J., Ris-Stalpers,C., Kuiper,G.G., Jenster,G.,
Berrevoets,C., Claassen,E., van Rooij,H.C., Trapman,J.,
Brinkmann,A.O. and Mulder,E.
TITLE A mutation in the ligand binding domain of the androgen receptor of
human LNCaP cells affects steroid binding characteristics and
response to anti-androgens
JOURNAL Biochem. Biophys. Res. Commun. 173 (2), 534-540 (1990)
MEDLINE 91083633
PUBMED 2260966
REMARK VARIANT LNCAP ALA-877.
REFERENCE 21 (residues 1 to 919)
AUTHORS Brown,T.R., Lubahn,D.B., Wilson,E.M., French,F.S., Migeon,C.J. and
Corden,J.L.
TITLE Functional characterization of naturally occurring mutant androgen
receptors from subjects with complete androgen insensitivity
JOURNAL Mol. Endocrinol. 4 (12), 1759-1772 (1990)
MEDLINE 91186983
PUBMED 2082179
REMARK VARIANTS CAIS CYS-774; GLN-831 AND MET-866.
REFERENCE 22 (residues 1 to 919)
AUTHORS Marcelli,M., Tilley,W.D., Zoppi,S., Griffin,J.E., Wilson,J.D. and
McPhaul,M.J.
TITLE Androgen resistance associated with a mutation of the androgen
receptor at amino acid 772 (Arg----Cys) results from a combination
of decreased messenger ribonucleic acid levels and impairment of
receptor function
JOURNAL J. Clin. Endocrinol. Metab. 73 (2), 318-325 (1991)
MEDLINE 91310758
PUBMED 1856263
REMARK VARIANT CYS-774.
REFERENCE 23 (residues 1 to 919)
AUTHORS Marcelli,M., Zoppi,S., Grino,P.B., Griffin,J.E., Wilson,J.D. and
McPhaul,M.J.
TITLE A mutation in the DNA-binding domain of the androgen receptor gene
causes complete testicular feminization in a patient with
receptor-positive androgen resistance
JOURNAL J. Clin. Invest. 87 (3), 1123-1126 (1991)
MEDLINE 91154385
PUBMED 1999491
REMARK VARIANT CAIS PRO-617.

REFERENCE 24 (residues 1 to 919)
AUTHORS McPhaul, M.J., Marcelli, M., Tilley, W.D., Griffin, J.E.,
Isidro-Gutierrez, R.F. and Wilson, J.D.
TITLE Molecular basis of androgen resistance in a family with a
qualitative abnormality of the androgen receptor and responsive to
high-dose androgen therapy
JOURNAL J. Clin. Invest. 87 (4), 1413-1421 (1991)
MEDLINE 91185626
PUBMED 2010552
REMARK VARIANT PAIS CYS-763.

REFERENCE 25 (residues 1 to 919)
AUTHORS Ris-Stalpers, C., Trifiro, M.A., Kuiper, G.G.J.M., Jenster, G.,
Romalo, G., Sai, T., van Rooij, H.C.J., Kaufman, M., Rosenfield, R.L.,
Liao, S., Schweikert, H.-U., Trapman, J., Pinsky, L. and Brinkmann, A.O.
TITLE Substitution of aspartic acid-686 by histidine or asparagine in the
human androgen receptor leads to a functionally inactive protein
with altered hormone-binding characteristics
JOURNAL Mol. Endocrinol. 5 (10), 1562-1569 (1991)
MEDLINE 92131007
PUBMED 1775137
REMARK VARIANTS CAIS ASN-695 AND HIS-695, AND SEQUENCE OF 629-723 FROM
N.A.

REFERENCE 26 (residues 1 to 919)
AUTHORS La Spada, A.R., Wilson, E.M., Lubahn, D.B., Harding, A.E. and
Fischbeck, K.H.
TITLE Androgen receptor gene mutations in X-linked spinal and bulbar
muscular atrophy
JOURNAL Nature 352 (6330), 77-79 (1991)
MEDLINE 91287825
PUBMED 2062380
REMARK VARIANTS SBMA IN POLY-GLN REGION.

REFERENCE 27 (residues 1 to 919)
AUTHORS Prior, L., Bordet, S., Trifiro, M.A., Mhatre, A., Kaufman, M.,
Pinsky, L., Wrogemann, K., Belsham, D.D., Pereira, F., Greenberg, C.R.,
Trapman, J., Brinkmann, A.O., Chang, C. and Liao, S.
TITLE Replacement of arginine 773 by cysteine or histidine in the human
androgen receptor causes complete androgen insensitivity with
different receptor phenotypes
JOURNAL Am. J. Hum. Genet. 51 (1), 143-155 (1992)
MEDLINE 92303560
PUBMED 1609793
REMARK VARIANTS CAIS CYS-774 AND HIS-774.

REFERENCE 28 (residues 1 to 919)
AUTHORS Saunders, P.T., Padayachi, T., Tincello, D.G., Shalet, S.M. and Wu, F.C.
TITLE Point mutations detected in the androgen receptor gene of three men
with partial androgen insensitivity syndrome
JOURNAL Clin. Endocrinol. (Oxf) 37 (3), 214-220 (1992)
MEDLINE 93047389
PUBMED 1424203
REMARK VARIANTS PAIS LYS-608 AND LEU-866.

REFERENCE 29 (residues 1 to 919)
AUTHORS Sweet, C.R., Behzadian, M.A. and McDonough, P.G.
TITLE A unique point mutation in the androgen receptor gene in a family
with complete androgen insensitivity syndrome
JOURNAL Fertil. Steril. 58 (4), 703-707 (1992)
MEDLINE 93050279
PUBMED 1426313
REMARK VARIANT CAIS THR-765.

REFERENCE 30 (residues 1 to 919)
AUTHORS Jakubiczka, S., Werder, E.A. and Wieacker, P.

TITLE Point mutation in the steroid-binding domain of the androgen receptor gene in a family with complete androgen insensitivity syndrome (CAIS)
JOURNAL Hum. Genet. 90 (3), 311-312 (1992)
MEDLINE 93138625
PUBMED 1487249
REMARK VARIANT CAIS VAL-749.
REFERENCE 31 (residues 1 to 919)
AUTHORS Batch,J.A., Williams,D.M., Davies,H.R., Brown,B.D., Evans,B.A., Hughes,I.A. and Patterson,M.N.

TITLE Androgen receptor gene mutations identified by SSCP in fourteen subjects with androgen insensitivity syndrome
JOURNAL Hum. Mol. Genet. 1 (7), 497-503 (1992)
MEDLINE 93338440
PUBMED 1307250
REMARK VARIANTS CAIS, AND VARIANTS PAIS.
REFERENCE 32 (residues 1 to 919)
AUTHORS Nakao,R., Haji,M., Yanase,T., Ogo,A., Takayanagi,R., Katsube,T., Fukumaki,Y. and Nawata,H.

TITLE A single amino acid substitution (Met786---Val) in the steroid-binding domain of human androgen receptor leads to complete androgen insensitivity syndrome
JOURNAL J. Clin. Endocrinol. Metab. 74 (5), 1152-1157 (1992)
MEDLINE 92235226
PUBMED 1569163
REMARK VARIANT CAIS VAL-787.
REFERENCE 33 (residues 1 to 919)
AUTHORS Wilson,C.M., Griffin,J.E., Wilson,J.D., Marcelli,M., Zoppi,S. and McPhaul,M.J.

TITLE Immunoreactive androgen receptor expression in subjects with androgen resistance
JOURNAL J. Clin. Endocrinol. Metab. 75 (6), 1474-1478 (1992)
MEDLINE 93100381
PUBMED 1464650
REMARK VARIANTS CAIS ARG-741 AND CYS-834.
REFERENCE 34 (residues 1 to 919)
AUTHORS McPhaul,M.J., Marcelli,M., Zoppi,S., Wilson,C.M., Griffin,J.E. and Wilson,J.D.

TITLE Mutations in the ligand-binding domain of the androgen receptor gene cluster in two regions of the gene
JOURNAL J. Clin. Invest. 90 (5), 2097-2101 (1992)
MEDLINE 93055453
PUBMED 1430233
REMARK VARIANTS CAIS, AND VARIANTS PAIS.
REFERENCE 35 (residues 1 to 919)
AUTHORS Veldscholte,J., Berrevoets,C.A., Ris-Stalpers,C., Kuiper,G.G., Jenster,G., Trapman,J., Brinkmann,A.O. and Mulder,E.

TITLE The androgen receptor in LNCaP cells contains a mutation in the ligand binding domain which affects steroid binding characteristics and response to antiandrogens
JOURNAL J. Steroid Biochem. Mol. Biol. 41 (3-8), 665-669 (1992)
MEDLINE 92222955
PUBMED 1562539
REMARK VARIANT PROSTATE CANCER ALA-877.
REFERENCE 36 (residues 1 to 919)
AUTHORS Zoppi,S., Marcelli,M., Deslypere,J.P., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.

TITLE Amino acid substitutions in the DNA-binding domain of the human androgen receptor are a frequent cause of receptor-binding positive androgen resistance

JOURNAL Mol. Endocrinol. 6 (3), 409-415 (1992)
MEDLINE 92261595
PUBMED 1316540
REMARK VARIANTS CAIS TYR-559 AND ARG-576, AND VARIANTS PAIS GLY-597 AND PRO-617.

REFERENCE 37 (residues 1 to 919)
AUTHORS De Bellis,A., Quigley,C.A., Cariello,N.F., el-Awady,M.K., Sar,M., Lane,M.V., Wilson,E.M. and French,F.S.
TITLE Single base mutations in the human androgen receptor gene causing complete androgen insensitivity: rapid detection by a modified denaturing gradient gel electrophoresis technique

JOURNAL Mol. Endocrinol. 6 (11), 1909-1920 (1992)
MEDLINE 93125565
PUBMED 1480178
REMARK VARIANTS CAIS SER-705; VAL-749; PHE-759; HIS-774; CYS-855 AND GLY-864.

REFERENCE 38 (residues 1 to 919)
AUTHORS Wooster,R., Mangion,J., Eeles,R., Smith,S., Dowsett,M., Averill,D., Barrett-Lee,P., Easton,D.F., Ponder,B.A. and Stratton,M.R.
TITLE A germline mutation in the androgen receptor gene in two brothers with breast cancer and Reifenstein syndrome

JOURNAL Nat. Genet. 2 (2), 132-134 (1992)
MEDLINE 93265137
PUBMED 1303262
REMARK VARIANT PAIS/BREAST CANCER GLN-607.

REFERENCE 39 (residues 1 to 919)
AUTHORS Newmark,J.R., Hardy,D.O., Tonb,D.C., Carter,B.S., Epstein,J.I., Isaacs,W.B., Brown,T.R. and Barrack,E.R.
TITLE Androgen receptor gene mutations in human prostate cancer

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 89 (14), 6319-6323 (1992)
MEDLINE 92335289
PUBMED 1631125
REMARK VARIANT MET-730.

REFERENCE 40 (residues 1 to 919)
AUTHORS Macke,J.P., Hu,N., Hu,S., Bailey,M., King,V.L., Brown,T., Hamer,D. and Nathans,J.
TITLE Sequence variation in the androgen receptor gene is not a common determinant of male sexual orientation

JOURNAL Am. J. Hum. Genet. 53 (4), 844-852 (1993)
MEDLINE 94027050
PUBMED 8213813
REMARK VARIANTS ARG-205 AND ASP-793.

REFERENCE 41 (residues 1 to 919)
AUTHORS Lumbroso,S., Lobaccaro,J.M., Belon,C., Martin,D., Chaussain,J.L. and Sultan,C.
TITLE A new mutation within the deoxyribonucleic acid-binding domain of the androgen receptor gene in a family with complete androgen insensitivity syndrome

JOURNAL Fertil. Steril. 60 (5), 814-819 (1993)
MEDLINE 94039857
PUBMED 8224266
REMARK VARIANT CAIS PHE-581.

REFERENCE 42 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Lumbroso,S., Ktari,R., Dumas,R. and Sultan,C.
TITLE An exonic point mutation creates a MaeIII site in the androgen receptor gene of a family with complete androgen insensitivity syndrome

JOURNAL Hum. Mol. Genet. 2 (7), 1041-1043 (1993)
MEDLINE 93372806
PUBMED 8103398

REMARK VARIANT CAIS VAL-754.
REFERENCE 43 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Lumbroso,S., Belon,C., Galtier-Dereure,F.,
Bringer,J., Lesimple,T., Namer,M., Cutuli,B.F., Pujol,H. and
Sultan,C.
TITLE Androgen receptor gene mutation in male breast cancer
JOURNAL Hum. Mol. Genet. 2 (11), 1799-1802 (1993)
MEDLINE 94108428
PUBMED 8281139
REMARK VARIANT PAIS/BREAST CANCER LYS-608.
REFERENCE 44 (residues 1 to 919)
AUTHORS Adeyemo,O., Kallio,P.J., Palvimo,J.J., Kontula,K. and Janne,O.A.
TITLE A single-base substitution in exon 6 of the androgen receptor gene
causing complete androgen insensitivity: the mutated receptor fails
to transactivate but binds to DNA in vitro
JOURNAL Hum. Mol. Genet. 2 (11), 1809-1812 (1993)
MEDLINE 94108430
PUBMED 8281140
REMARK VARIANT CAIS ARG-807.
REFERENCE 45 (residues 1 to 919)
AUTHORS Nakao,R., Yanase,T., Sakai,Y., Haji,M. and Nawata,H.
TITLE A single amino acid substitution (gly743 --> val) in the
steroid-binding domain of the human androgen receptor leads to
Reifenstein syndrome
JOURNAL J. Clin. Endocrinol. Metab. 77 (1), 103-107 (1993)
MEDLINE 93315568
PUBMED 8325932
REMARK VARIANT PAIS VAL-743.
REFERENCE 46 (residues 1 to 919)
AUTHORS Hiort,O., Huang,Q., Sinnecker,G.H., Sadeghi-Nejad,A., Kruse,K.,
Wolfe,H.J. and Yandell,D.W.
TITLE Single strand conformation polymorphism analysis of androgen
receptor gene mutations in patients with androgen insensitivity
syndromes: application for diagnosis, genetic counseling, and
therapy
JOURNAL J. Clin. Endocrinol. Metab. 77 (1), 262-266 (1993)
MEDLINE 93315600
PUBMED 8325950
REMARK VARIANTS CAIS LYS-681 AND THR-842, AND VARIANTS PAIS HIS-840 AND
LEU-866.
REFERENCE 47 (residues 1 to 919)
AUTHORS Batch,J.A., Evans,B.A., Hughes,I.A. and Patterson,M.N.
TITLE Mutations of the androgen receptor gene identified in perineal
hypospadias
JOURNAL J. Med. Genet. 30 (3), 198-201 (1993)
MEDLINE 93233131
PUBMED 8097257
REMARK VARIANTS PAIS HIS-855 AND MET-869.
REFERENCE 48 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Lumbroso,S., Berta,P., Chaussain,J.L. and Sultan,C.
TITLE Complete androgen insensitivity syndrome associated with a de novo
mutation of the androgen receptor gene detected by single strand
conformation polymorphism
JOURNAL J. Steroid Biochem. Mol. Biol. 44 (3), 211-216 (1993)
MEDLINE 93213715
PUBMED 8096390
REMARK VARIANT CAIS VAL-743.
REFERENCE 49 (residues 1 to 919)
AUTHORS Suzuki,H., Sato,N., Watabe,Y., Masai,M., Seino,S. and Shimazaki,J.
TITLE Androgen receptor gene mutations in human prostate cancer

JOURNAL J. Steroid Biochem. Mol. Biol. 46 (6), 759-765 (1993)
MEDLINE 94100129
PUBMED 8274409
REMARK VARIANTS PROSTATE CANCER HIS-701 AND ALA-877.
REFERENCE 50 (residues 1 to 919)
AUTHORS Kazemi-Esfarjani, P., Beitel, L.K., Trifiro, M., Kaufman, M.,
Rennie, P., Sheppard, P., Matusik, R. and Pinsky, L.
TITLE Substitution of valine-865 by methionine or leucine in the human
androgen receptor causes complete or partial androgen
insensitivity, respectively with distinct androgen receptor
phenotypes

JOURNAL Mol. Endocrinol. 7 (1), 37-46 (1993)
MEDLINE 93188862
PUBMED 8446106
REMARK VARIANT CAIS MET-866, AND VARIANT PAIS LEU-866.
REFERENCE 51 (residues 1 to 919)
AUTHORS Mowszowicz, I., Lee, H.J., Chen, H.T., Mestayer, C., Portois, M.C.,
Cabrol, S., Mauvais-Jarvis, P. and Chang, C.
TITLE A point mutation in the second zinc finger of the DNA-binding
domain of the androgen receptor gene causes complete androgen
insensitivity in two siblings with receptor-positive androgen
resistance

JOURNAL Mol. Endocrinol. 7 (7), 861-869 (1993)
MEDLINE 94019395
PUBMED 8413310
REMARK VARIANT CAIS HIS-615.
REFERENCE 52 (residues 1 to 919)
AUTHORS Culig, Z., Hobisch, A., Cronauer, M.V., Cato, A.C., Hittmair, A.,
Radmayr, C., Eberle, J., Bartsch, G. and Klocker, H.
TITLE Mutant androgen receptor detected in an advanced-stage prostatic
carcinoma is activated by adrenal androgens and progesterone

JOURNAL Mol. Endocrinol. 7 (12), 1541-1550 (1993)
MEDLINE 94195311
PUBMED 8145761
REMARK VARIANT PROSTATE CANCER MET-715.
REFERENCE 53 (residues 1 to 919)
AUTHORS Lobaccaro, J.-M., Lumbroso, S., Belon, C., Chaussain, J.L.,
Toublanc, J.E., Leheup, B. and Sultan, C.
TITLE Androgen receptor (AR) gene mutations in 6 families with androgen
insensitivity syndrome (Abstract #114)

JOURNAL Pediatr. Res. Suppl. 33, S22-S22 (1993)
REMARK VARIANTS CAIS PHE-581; VAL-743; VAL-754; GLU-767 AND CYS-855.
REFERENCE 54 (residues 1 to 919)
AUTHORS Castagnaro, M., Yandell, D.W., Dockhorn-Dworniczak, B., Wolfe, H.J. and
Poremba, C.
TITLE Androgen receptor gene mutations and p53 gene analysis in advanced
prostate cancer

JOURNAL Verh Dtsch Ges Pathol 77, 119-123 (1993)
MEDLINE 94189145
PUBMED 7511268
REMARK VARIANTS PROSTATE CANCER LEU-340 AND GLU-798.
REFERENCE 55 (residues 1 to 919)
AUTHORS Schoenberg, M.P., Hakimi, J.M., Wang, S., Bova, G.S., Epstein, J.I.,
Fischbeck, K.H., Isaacs, W.B., Walsh, P.C. and Barrack, E.R.
TITLE Microsatellite mutation (CAG24-->18) in the androgen receptor gene
in human prostate cancer

JOURNAL Biochem. Biophys. Res. Commun. 198 (1), 74-80 (1994)
MEDLINE 94121667
PUBMED 8292051
REMARK VARIANT PROSTATE CANCER IN POLY-GLN REGION.

REFERENCE 56 (residues 1 to 919)
AUTHORS Gaddipati,J.P., McLeod,D.G., Heidenberg,H.B., Sesterhenn,I.A.,
Finger,M.J., Moul,J.W. and Srivastava,S.
TITLE Frequent detection of codon 877 mutation in the androgen receptor
gene in advanced prostate cancers
JOURNAL Cancer Res. 54 (11), 2861-2864 (1994)
MEDLINE 94243798
PUBMED 8187068
REMARK VARIANT PROSTATE CANCER ALA-877.

REFERENCE 57 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Belon,C., Lumbroso,S., Olewniczack,G.,
Carre-Pigeon,F., Job,J.C., Chaussain,J.L., Toublanc,J.E. and
Sultan,C.
TITLE Molecular prenatal diagnosis of partial androgen insensitivity
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PUBMED 7910529
REMARK VARIANT PAIS TRP-568.

REFERENCE 58 (residues 1 to 919)
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PUBMED 8205256
REMARK VARIANT PAIS HIS-840.

REFERENCE 60 (residues 1 to 919)
AUTHORS Hiort,O., Klauber,G., Cendron,M., Sinnecker,G.H., Keim,L.,
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REMARK VARIANT PAIS VAL-870.

REFERENCE 61 (residues 1 to 919)
AUTHORS Schwartz,M., Skovby,F., Mueller,J., Nielsen,O. and Skakkebaek,N.E.
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REFERENCE 62 (residues 1 to 919)
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REMARK VARIANTS CAIS PHE-582 DEL; ARG-615 DEL AND HIS-615.
REFERENCE 63 (residues 1 to 919)
AUTHORS Hiort,O., Wodtke,A., Struve,D., Zollner,A. and Sinnecker,G.H.
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PUBMED 7981687
REMARK VARIANTS PAIS SER-582; TYR-604; ALA-708; LEU-754 AND HIS-771, AND VARIANT CAIS TRP-779.
REFERENCE 64 (residues 1 to 919)
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PUBMED 7981689
REMARK VARIANT CAIS PHE-601.
REFERENCE 65 (residues 1 to 919)
AUTHORS De Bellis,A., Quigley,C.A., Marschke,K.B., el-Awady,M.K., Lane,M.V., Smith,E.P., Sar,M., Wilson,E.M. and French,F.S.
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REMARK VARIANTS PAIS ARG-616; HIS-840 AND MET-889.
REFERENCE 66 (residues 1 to 919)
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REMARK VARIANT MAIS PHE-790.
REFERENCE 67 (residues 1 to 919)
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AUTHORS Marcelli,M., Zoppi,S., Wilson,C.M., Griffin,J.E. and McPhaul,M.J.
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REMARK VARIANTS CAIS, AND VARIANTS PAIS.
REFERENCE 69 (residues 1 to 919)
AUTHORS Yong,E.L., Ng,S.C., Roy,A.C., Yun,G. and Ratnam,S.S.
TITLE Pregnancy after hormonal correction of severe spermatogenic defect due to mutation in androgen receptor gene
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REMARK VARIANT MAIS LYS-727.
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AUTHORS Ris-Stalpers,C., Hoogenboezem,T., Sleddens,H.F.B.M., Verleun-Mooijman,M.C.T., Degenhart,H.J., Drop,S.L.S., Halley,D.J.J., Oosterwijk,J.C., Hodgins,M.B., Trapman,J. and Brinkmann,A.O.
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AUTHORS Imai,A., Ohno,T., Iida,K., Ohsuye,K., Okano,Y. and Tamaya,T.
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TITLE Prevalence of androgen receptor gene mutations in latent prostatic carcinomas from Japanese men
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REMARK VARIANTS PROSTATE CANCER.
REFERENCE 73 (residues 1 to 919)
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REFERENCE 74 (residues 1 to 919)
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THR-737; HIS-774 AND GLU-798.

REFERENCE 75 (residues 1 to 919)

AUTHORS Quigley,C.A., De Bellis,A., Marschke,K.B., el-Awady,M.K.,
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REMARK VARIANTS CAIS LEU-831 AND GLN-831.

REFERENCE 77 (residues 1 to 919)

AUTHORS Belsham,D.D., Pereira,F., Greenberg,C.R., Liao,S. and Wrogemann,K.

TITLE Leu-676-Pro mutation of the androgen receptor causes complete
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REMARK VARIANT CAIS PRO-677.

REFERENCE 78 (residues 1 to 919)

AUTHORS Murono,K., Mendonca,B.B., Arnhold,I.J., Rigon,A.C., Migeon,C.J. and
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TITLE Human androgen insensitivity due to point mutations encoding amino
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CYS-855.

REFERENCE 79 (residues 1 to 919)

AUTHORS Peterziel,H., Culig,Z., Stober,J., Hobisch,A., Radmayr,C.,
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REMARK VARIANT PROSTATE CANCER MET-730.

REFERENCE 80 (residues 1 to 919)

AUTHORS Allera,A., Herbst,M.A., Griffin,J.E., Wilson,J.D., Schweikert,H.U.
and McPhaul,M.J.

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REMARK VARIANT PAIS VAL-568.

REFERENCE 81 (residues 1 to 919)

AUTHORS Elo,J.P., Kvist,L., Leinonen,K., Isomaa,V., Henttu,P.,
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PUBMED 8530589
REMARK VARIANT PROSTATE CANCER LEU-726.
REFERENCE 82 (residues 1 to 919)
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REMARK VARIANT PAIS THR-596.
REFERENCE 83 (residues 1 to 919)
AUTHORS Taplin,M.E., Bubley,G.J., Shuster,T.D., Frantz,M.E., Spooner,A.E., Ogata,G.K., Keer,H.N. and Balk,S.P.
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REMARK VARIANTS PROSTATE CANCER.
REFERENCE 84 (residues 1 to 919)
AUTHORS Hiort,O., Sinnecker,G.H., Holterhus,P.M., Nitsche,E.M. and Kruse,K.
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JOURNAL Am. J. Med. Genet. 63 (1), 218-222 (1996)
MEDLINE 96298278
PUBMED 8723113
REMARK VARIANTS CAIS AND PAIS.
REFERENCE 85 (residues 1 to 919)
AUTHORS Tilley,W.D., Buchanan,G., Hickey,T.E. and Bentel,J.M.
TITLE Mutations in the androgen receptor gene are associated with progression of human prostate cancer to androgen independence
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REMARK VARIANTS PROSTATE CANCER.
REFERENCE 86 (residues 1 to 919)
AUTHORS Weidemann,W., Linck,B., Haupt,H., Mentrup,B., Romalo,G., Stockklauser,K., Brinkmann,A.O., Schweikert,H.U. and Spindler,K.D.
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MEDLINE 97191382
PUBMED 9039340
REMARK VARIANTS PAIS GLN-607; THR-610; LEU-754; HIS-840; THR-842 AND HIS-855, AND VARIANT CAIS MET-866.
REFERENCE 87 (residues 1 to 919)
AUTHORS Malmgren,H., Gustavsson,J., Tuvemo,T. and Dahl,N.
TITLE Rapid detection of a mutation hot-spot in the human androgen receptor
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REMARK VARIANT CAIS CYS-855.
REFERENCE 88 (residues 1 to 919)
AUTHORS Bevan,C.L., Brown,B.B., Davies,H.R., Evans,B.A., Hughes,I.A. and Patterson,M.N.
TITLE Functional analysis of six androgen receptor mutations identified in patients with partial androgen insensitivity syndrome
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MEDLINE 96422266
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REMARK VARIANTS PAIS ILE-742; ILE-780; GLU-798; CYS-840; HIS-855 AND MET-869.

REFERENCE 89 (residues 1 to 919)
AUTHORS Choong,C.S., Sturm,M.J., Strophair,J.A., McCulloch,R.K., Tilley,W.D., Leedman,P.J. and Hurley,D.M.
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JOURNAL J. Clin. Endocrinol. Metab. 81 (1), 236-243 (1996)
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PUBMED 8550758
REMARK VARIANT PAIS ARG-909.

REFERENCE 90 (residues 1 to 919)
AUTHORS Lumbroso,S., Lobaccaro,J.M., Georget,V., Leger,J., Poujol,N., Terouanne,B., Evain-Brion,D., Czernichow,P. and Sultan,C.
TITLE A novel substitution (Leu707Arg) in exon 4 of the androgen receptor gene causes complete androgen resistance
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MEDLINE 96210890
PUBMED 8626869
REMARK VARIANT CAIS ARG-707.

REFERENCE 91 (residues 1 to 919)
AUTHORS Rodien,P., Mebarki,F., Mowszowicz,I., Chaussain,J.L., Young,J., Morel,Y. and Schaison,G.
TITLE Different phenotypes in a family with androgen insensitivity caused by the same M780I point mutation in the androgen receptor gene
JOURNAL J. Clin. Endocrinol. Metab. 81 (8), 2994-2998 (1996)
MEDLINE 96320094
PUBMED 8768864
REMARK VARIANT PAIS/CAIS ILE-780.

REFERENCE 92 (residues 1 to 919)
AUTHORS Choong,C.S., Quigley,C.A., French,F.S. and Wilson,E.M.
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MEDLINE 96420610
PUBMED 8823308
REMARK VARIANT PAIS LYS-2.

REFERENCE 93 (residues 1 to 919)
AUTHORS Bruggenwirth,H.T., Boehmer,A.L., Verleun-Mooijman,M.C., Hoogenboezem,T., Kleijer,W.J., Otten,B.J., Trapman,J. and Brinkmann,A.O.
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PUBMED 8918984
REMARK VARIANT CAIS ASP-573.

REFERENCE 94 (residues 1 to 919)
AUTHORS Sutherland,R.W., Wiener,J.S., Hicks,J.P., Marcelli,M., Gonzales,E.T. Jr., Roth,D.R. and Lamb,D.J.
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MEDLINE 96289452
PUBMED 8683794

REMARK VARIANT MAIS SER-548.
REFERENCE 95 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Poujol,N., Chiche,L., Lumbroso,S., Brown,T.R. and Sultan,C.
TITLE Molecular modeling and in vitro investigations of the human androgen receptor DNA-binding domain: application for the study of two mutations
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REMARK VARIANT CAIS PRO-616.
REFERENCE 96 (residues 1 to 919)
AUTHORS Imasaki,K., Okabe,T., Murakami,H., Tanaka,Y., Haji,M., Takayanagi,R. and Nawata,H.
TITLE Androgen insensitivity syndrome due to new mutations in the DNA-binding domain of the androgen receptor
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REMARK VARIANT CAIS PHE-579, AND VARIANT PAIS TYR-582.
REFERENCE 97 (residues 1 to 919)
AUTHORS Evans,B.A., Harper,M.E., Daniells,C.E., Watts,C.E., Matenhelia,S., Green,J. and Griffiths,K.
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PUBMED 8628719

REMARK VARIANT PROSTATE CANCER GLU-798.
REFERENCE 98 (residues 1 to 919)
AUTHORS Suzuki,H., Akakura,K., Komiya,A., Aida,S., Akimoto,S. and Shimazaki,J.
TITLE Codon 877 mutation in the androgen receptor gene in advanced prostate cancer: relation to antiandrogen withdrawal syndrome
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MEDLINE 96424622
PUBMED 8827083

REMARK VARIANT PROSTATE CANCER ALA-877.
REFERENCE 99 (residues 1 to 919)
AUTHORS Boehmer,A.L., Brinkmann,A.O., Niermeijer,M.F., Bakker,L., Halley,D.J. and Drop,S.L.
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JOURNAL Am. J. Hum. Genet. 60 (4), 1003-1006 (1997)
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PUBMED 9106550

REMARK VARIANT PAIS HIS-855.
REFERENCE 100 (residues 1 to 919)
AUTHORS Koivisto,P., Kononen,J., Palmberg,C., Tammela,T., Hyytinen,E., Isola,J., Trapman,J., Cleutjens,K., Noordzij,A., Visakorpi,T. and Kallioniemi,O.-P.
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JOURNAL Cancer Res. 57 (2), 314-319 (1997)
MEDLINE 97153285
PUBMED 9000575

REMARK VARIANT PROSTATE CANCER ALA-683.
REFERENCE 101 (residues 1 to 919)

AUTHORS Tincello,D.G., Saunders,P.T., Hodgins,M.B., Simpson,N.B.,
Edwards,C.R., Hargreaves,T.B. and Wu,F.C.

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partial androgen insensitivity syndrome

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PUBMED 9196614

REMARK VARIANTS PAIS LYS-608 AND GLY-772.

REFERENCE 102 (residues 1 to 919)

AUTHORS Essawi,M., Gad,Y.Z., el-Rouby,O., Temtamy,S.A., Sabour,Y.A. and
el-Awady,M.K.

TITLE Molecular analysis of androgen resistance syndromes in Egyptian
patients

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PUBMED 9160185

REMARK VARIANT PAIS MET-889.

REFERENCE 103 (residues 1 to 919)

AUTHORS Sinnecker,G.H., Hiort,O., Nitsche,E.M., Holterhus,P.M. and Kruse,K.

TITLE Functional assessment and clinical classification of androgen
sensitivity in patients with mutations of the androgen receptor
gene. German Collaborative Intersex Study Group

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REMARK VARIANT CAIS TRP-779.

REFERENCE 104 (residues 1 to 919)

AUTHORS Jakubiczka,S., Nedel,S., Werder,E.A., Schleiermacher,E., Theile,U.,
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TITLE Mutations of the androgen receptor gene in patients with complete
androgen insensitivity

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REMARK VARIANTS CAIS VAL-749; CYS-774; ILE-780 AND SER-794.

REFERENCE 105 (residues 1 to 919)

AUTHORS Watanabe,M., Ushijima,T., Shiraishi,T., Yatani,R., Shimazaki,J.,
Kotake,T., Sugimura,T. and Nagao,M.

TITLE Genetic alterations of androgen receptor gene in Japanese human
prostate cancer

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REMARK VARIANTS PROSTATE CANCER IN POLY-GLN REGION; HIS-701 AND ARG-910.

REFERENCE 106 (residues 1 to 919)

AUTHORS Wang,C. and Uchida,T.

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REMARK VARIANT PROSTATE CANCER GLN-629.

REFERENCE 107 (residues 1 to 919)

AUTHORS Komori,S., Sakata,K., Tanaka,H., Shima,H. and Koyama,K.

TITLE DNA analysis of the androgen receptor gene in two cases with
complete androgen insensitivity syndrome

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REMARK VARIANTS CAIS ARG-194 AND CYS-855.

REFERENCE 108 (residues 1 to 919)

AUTHORS Albers,N., Ulrichs,C., Gluer,S., Hiort,O., Sinnecker,G.H.,

TITLE Mildenerger,H. and Brodehl,J.
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MEDLINE 97469997
PUBMED 9329414
REMARK VARIANTS PAIS ALA-708 AND GLY-870.
REFERENCE 109 (residues 1 to 919)
AUTHORS Ko,T.M., Yang,Y.S., Wu,M.Y., Kao,C.H., Hsu,P.M., Chuang,S.M. and Lee,T.Y.

TITLE Complete androgen insensitivity syndrome. Molecular characterization in two Chinese women
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PUBMED 9252933
REMARK VARIANTS CAIS ASN-732 AND THR-765.
REFERENCE 110 (residues 1 to 919)
AUTHORS Bevan,C.L., Hughes,I.A. and Patterson,M.N.

TITLE Wide variation in androgen receptor dysfunction in complete androgen insensitivity syndrome
JOURNAL J. Steroid Biochem. Mol. Biol. 61 (1-2), 19-26 (1997)
MEDLINE 97466866
PUBMED 9328206
REMARK VARIANTS CAIS ASP-750; PHE-762; THR-765; ASN-864 AND PHE-907.
REFERENCE 111 (residues 1 to 919)
AUTHORS Radmayr,C., Culig,Z., Glatzl,J., Neuschmid-Kaspar,F., Bartsch,G. and Klocker,H.

TITLE Androgen receptor point mutations as the underlying molecular defect in 2 patients with androgen insensitivity syndrome
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MEDLINE 97445885
PUBMED 9302173
REMARK VARIANT PAIS GLY-703, AND VARIANT CAIS LEU-916.
REFERENCE 112 (residues 1 to 919)
AUTHORS Komori,S., Kasumi,H., Sakata,K., Tanaka,H., Hamada,K. and Koyama,K.

TITLE Molecular analysis of the androgen receptor gene in 4 patients with complete androgen insensitivity
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REMARK VARIANTS CAIS CYS-571; GLN-752 AND CYS-774.
REFERENCE 113 (residues 1 to 919)
AUTHORS Cabral,D.F., Maciel-Guerra,A.T. and Hackel,C.

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PUBMED 9698822
REMARK VARIANTS CAIS HIS-615 AND GLN-752.
REFERENCE 114 (residues 1 to 919)
AUTHORS Wang,Q., Ghadessy,F.J. and Yong,E.L.

TITLE Analysis of the transactivation domain of the androgen receptor in patients with male infertility
JOURNAL Clin. Genet. 54 (3), 185-192 (1998)
MEDLINE 99002768
PUBMED 9788719
REMARK VARIANT MAIS ARG-214.
REFERENCE 115 (residues 1 to 919)
AUTHORS Tanaka,H., Komori,S., Sakata,K., Shima,H. and Koyama,K.

TITLE One additional mutation at exon A amplifies thermolability of

androgen receptor in a case with complete androgen insensitivity syndrome

JOURNAL Gynecol Endocrinol 12 (2), 75-82 (1998)
MEDLINE 98273339
PUBMED 9610419
REMARK VARIANTS CAIS PRO-255 AND ALA-820.
REFERENCE 116 (residues 1 to 919)
AUTHORS Lundberg Giwerzman, Y., Nikoshkov, A., Lindsten, K., Bystroem, B., Pousette, A., Chibalin, A.V., Arvidsson, S., Tiulpakov, A., Semitcheva, T.V., Peterkova, V., Hagenfeldt, K., Ritzen, E.M. and Wedell, A.

TITLE Functional characterisation of mutations in the ligand-binding domain of the androgen receptor gene in patients with androgen insensitivity syndrome

JOURNAL Hum. Genet. 103 (4), 529-531 (1998)
MEDLINE 99072324
PUBMED 9856504
REMARK VARIANTS CAIS THR-765; TYR-784 AND THR-895, AND VARIANT PAIS GLY-840.

REFERENCE 117 (residues 1 to 919)
AUTHORS Doerk, T., Schnieders, F., Jakubiczka, S., Wieacker, P., Schroeder-Kurth, T. and Schmidtke, J.

TITLE A new missense substitution at a mutational hot spot of the androgen receptor in siblings with complete androgen insensitivity syndrome

JOURNAL Hum. Mutat. 11, 337-339 (1998)
REMARK VARIANT CAIS VAL-695.

REFERENCE 118 (residues 1 to 919)
AUTHORS Nordenskjöld, A. and Soederhaell, S.

TITLE An androgen receptor gene mutation (A645D) in a boy with a normal phenotype

JOURNAL Hum. Mutat. 11, 339-339 (1998)
REMARK VARIANT ASP-645.

REFERENCE 119 (residues 1 to 919)
AUTHORS Knoke, I., Jakubiczka, S., Rohrer, T., Hanimann, B., Werder, E.A. and Wieacker, P.

TITLE Single amino acid substitution in the hormone-binding domain of the androgen receptor in a family with complete androgen insensitivity syndrome (CAIS)

JOURNAL Hum. Mutat. 12, 220-220 (1998)
REMARK VARIANT CAIS LEU-892.

REFERENCE 120 (residues 1 to 919)
AUTHORS Weidemann, W., Peters, B., Romalo, G., Spindler, K.D. and Schweikert, H.U.

TITLE Response to androgen treatment in a patient with partial androgen insensitivity and a mutation in the deoxyribonucleic acid-binding domain of the androgen receptor

JOURNAL J. Clin. Endocrinol. Metab. 83 (4), 1173-1176 (1998)
MEDLINE 98202136
PUBMED 9543136
REMARK VARIANT PAIS GLN-607.

REFERENCE 121 (residues 1 to 919)
AUTHORS Georget, V., Terouanne, B., Lumbroso, S., Nicolas, J.C. and Sultan, C.

TITLE Trafficking of androgen receptor mutants fused to green fluorescent protein: a new investigation of partial androgen insensitivity syndrome

JOURNAL J. Clin. Endocrinol. Metab. 83 (10), 3597-3603 (1998)
MEDLINE 98439607
PUBMED 9768671
REMARK VARIANTS PAIS VAL-743 AND CYS-840.

REFERENCE 122 (residues 1 to 919)
AUTHORS Wang,Q., Ghadessy,F.J., Trounson,A., de Kretser,D., McLachlan,R.,
Ng,S.C. and Yong,E.L.
TITLE Azoospermia associated with a mutation in the ligand-binding domain
of an androgen receptor displaying normal ligand binding, but
defective trans-activation
JOURNAL J. Clin. Endocrinol. Metab. 83 (12), 4303-4309 (1998)
MEDLINE 99067093
PUBMED 9851768
REMARK VARIANT MAIS GLU-798.

REFERENCE 123 (residues 1 to 919)
AUTHORS Hiort,O., Sinnecker,G.H., Holterhus,P.M., Nitsche,E.M. and Kruse,K.
TITLE Inherited and de novo androgen receptor gene mutations:
investigation of single-case families
JOURNAL J. Pediatr. 132 (6), 939-943 (1998)
MEDLINE 98291052
PUBMED 9627582
REMARK VARIANTS CAIS AND PAIS.

REFERENCE 124 (residues 1 to 919)
AUTHORS Yong,E.L., Tut,T.G., Ghadessy,F.J., Prins,G. and Ratnam,S.S.
TITLE Partial androgen insensitivity and correlations with the predicted
three dimensional structure of the androgen receptor ligand-binding
domain
JOURNAL Mol. Cell. Endocrinol. 137 (1), 41-50 (1998)
MEDLINE 98268743
PUBMED 9607727
REMARK VARIANT PAIS THR-758.

REFERENCE 125 (residues 1 to 919)
AUTHORS Knoke,I., Jakubiczka,S., Lehnert,H. and Wieacker,P.
TITLE A new point mutation of the androgen receptor gene in a patient
with partial androgen resistance and severe oligozoospermia
JOURNAL Andrologia 31 (4), 199-201 (1999)
MEDLINE 99399430
PUBMED 10470409
REMARK VARIANT PAIS LEU-911.

REFERENCE 126 (residues 1 to 919)
AUTHORS Taplin,M.E., Bubley,G.J., Ko,Y.J., Small,E.J., Upton,M.,
Rajeshkumar,B. and Balk,S.P.
TITLE Selection for androgen receptor mutations in prostate cancers
treated with androgen antagonist
JOURNAL Cancer Res. 59 (11), 2511-2515 (1999)
MEDLINE 99290631
PUBMED 10363963
REMARK VARIANTS PROSTATE CANCER ALA-877 AND ASN-890.

REFERENCE 127 (residues 1 to 919)
AUTHORS Melo,K.F.S., Latronico,A.C., Costa,E.M.F., Billerbeck,A.E.C.,
Mendonca,B.B. and Arnhold,I.J.P.
TITLE A novel point mutation (R840S) in the androgen receptor in a
Brazilian family with partial androgen insensitivity syndrome
JOURNAL Hum. Mutat. 14, 353-353 (1999)
REMARK VARIANT PAIS SER-840.

REFERENCE 128 (residues 1 to 919)
AUTHORS Gottlieb,B., Vasiliou,D.M., Lumbroso,R., Beitel,L.K., Pinsky,L. and
Trifiro,M.A.
TITLE Analysis of exon 1 mutations in the androgen receptor gene
JOURNAL Hum. Mutat. 14 (6), 527-539 (1999)
MEDLINE 20040031
PUBMED 10571951
REMARK VARIANTS CAIS ARG-390 AND ARG-443.

REFERENCE 129 (residues 1 to 919)

AUTHORS Chen, C.P., Chern, S.R., Wang, T.Y., Wang, W., Wang, K.L. and Jeng, C.J.
TITLE Androgen receptor gene mutations in 46,XY females with germ cell tumours
JOURNAL Hum. Reprod. 14 (3), 664-670 (1999)
MEDLINE [99236881](#)
PUBMED [10221692](#)
REMARK VARIANT PAIS GLN-607, AND VARIANT CAIS LYS-681.
REFERENCE 130 (residues 1 to 919)
AUTHORS Kanayama, H., Naroda, T., Inoue, Y., Kurokawa, Y. and Kagawa, S.
TITLE A case of complete testicular feminization: laparoscopic orchiectomy and analysis of androgen receptor gene mutation
JOURNAL Int J Urol 6 (6), 327-330 (1999)
MEDLINE [99332382](#)
PUBMED [10404311](#)
REMARK VARIANT CAIS LEU-892.
REFERENCE 131 (residues 1 to 919)
AUTHORS Shkolny, D.L., Beitel, L.K., Ginsberg, J., Pেকেles, G., Arbour, L., Pinsky, L. and Trifiro, M.A.
TITLE Discordant measures of androgen-binding kinetics in two mutant androgen receptors causing mild or partial androgen insensitivity, respectively
JOURNAL J. Clin. Endocrinol. Metab. 84 (2), 805-810 (1999)
MEDLINE [99145056](#)
PUBMED [10022458](#)
REMARK VARIANT PAIS ALA-772, AND VARIANT MAIS GLY-871.
REFERENCE 132 (residues 1 to 919)
AUTHORS Wallen, M.J., Linja, M., Kaartinen, K., Schleutker, J. and Visakorpi, T.
TITLE Androgen receptor gene mutations in hormone-refractory prostate cancer
JOURNAL J. Pathol. 189 (4), 559-563 (1999)
MEDLINE [20096941](#)
PUBMED [10629558](#)
REMARK VARIANTS PROSTATE CANCER IN POLY-GLN REGION AND ALA-683.
REFERENCE 133 (residues 1 to 919)
AUTHORS Zhao, X.Y., Boyle, B., Krishnan, A.V., Navone, N.M., Peehl, D.M. and Feldman, D.
TITLE Two mutations identified in the androgen receptor of the new human prostate cancer cell line MDA PCa 2a
JOURNAL J. Urol. 162 (6), 2192-2199 (1999)
MEDLINE [20034906](#)
PUBMED [10569618](#)
REMARK VARIANTS PROSTATE CANCER HIS-701 AND ALA-877.
REFERENCE 134 (residues 1 to 919)
AUTHORS Ong, Y.C., Wong, H.B., Adaikan, G. and Yong, E.L.
TITLE Directed pharmacological therapy of ambiguous genitalia due to an androgen receptor gene mutation
JOURNAL Lancet 354 (9188), 1444-1445 (1999)
MEDLINE [20009021](#)
PUBMED [10543676](#)
REMARK VARIANT PAIS THR-807.
REFERENCE 135 (residues 1 to 919)
AUTHORS Peters, I., Weidemann, W., Romalo, G., Knorr, D., Schweikert, H.U. and Spindler, K.D.
TITLE An androgen receptor mutation in the direct vicinity of the proposed C-terminal alpha-helix of the ligand binding domain containing the AF-2 transcriptional activating function core is associated with complete androgen insensitivity
JOURNAL Mol. Cell. Endocrinol. 148 (1-2), 47-53 (1999)
MEDLINE [99236959](#)
PUBMED [10221770](#)

REMARK VARIANT CAIS LEU-892.
REFERENCE 136 (residues 1 to 919)
AUTHORS Nazareth,L.V., Stenoien,D.L., Bingman,W.E. III, James,A.J., Wu,C., Zhang,Y., Edwards,D.P., Mancini,M., Marcelli,M., Lamb,D.J. and Weigel,N.L.
TITLE A C619Y mutation in the human androgen receptor causes inactivation and mislocalization of the receptor with concomitant sequestration of SRC-1 (steroid receptor coactivator 1)
JOURNAL Mol. Endocrinol. 13 (12), 2065-2075 (1999)
MEDLINE 20065641
PUBMED 10598582
REMARK VARIANT PROSTATE CANCER TYR-619.
REFERENCE 137 (residues 1 to 919)
AUTHORS Nazareth,L.V., Stenoien,D.L., Bingman,W.E. III, James,A.J., Wu,C., Zhang,Y., Edwards,D.P., Mancini,M., Marcelli,M., Lamb,D.J. and Weigel,N.L.
JOURNAL Mol. Endocrinol. 14, 544-544 (2000)
REMARK ERRATUM.
REFERENCE 138 (residues 1 to 919)
AUTHORS Holterhus,P.M., Wiebel,J., Sinnecker,G.H., Bruggenwirth,H.T., Sippell,W.G., Brinkmann,A.O., Kruse,K. and Hiort,O.
TITLE Clinical and molecular spectrum of somatic mosaicism in androgen insensitivity syndrome
JOURNAL Pediatr. Res. 46 (6), 684-690 (1999)
MEDLINE 20055969
PUBMED 10590024
REMARK VARIANT PAIS THR-596.
REFERENCE 139 (residues 1 to 919)
AUTHORS Yaegashi,N., Uehara,S., Senoo,M., Sato,J., Fujiwara,J., Funato,T., Sasaki,T. and Yajima,A.
TITLE Point mutations in the steroid-binding domain of the androgen receptor gene of five Japanese patients with androgen insensitivity syndrome
JOURNAL Tohoku J. Exp. Med. 187 (3), 263-272 (1999)
MEDLINE 99385665
PUBMED 10458483
REMARK VARIANT PAIS PHE-812, AND VARIANT CAIS GLN-831.
REFERENCE 140 (residues 1 to 919)
AUTHORS Nordenskjold,A., Friedman,E., Tapper-Persson,M., Soderhall,C., Leviav,A., Svensson,J. and Anvret,M.
TITLE Screening for mutations in candidate genes for hypospadias
JOURNAL Urol. Res. 27 (1), 49-55 (1999)
MEDLINE 99190574
PUBMED 10092153
REMARK VARIANTS PAIS THR-597 AND LEU-725.
REFERENCE 141 (residues 1 to 919)
AUTHORS Marcelli,M., Ittmann,M., Mariani,S., Sutherland,R.W., Nigam,R., Murthy,L., Zhao,Y., DiConcini,D., Puxeddu,E., Esen,A., Eastham,J., Weigel,N.L. and Lamb,D.J.
TITLE Androgen receptor mutations in prostate cancer
JOURNAL Cancer Res. 60 (4), 944-949 (2000)
MEDLINE 20168626
PUBMED 10706109
REMARK VARIANTS PROSTATE CANCER ALA-575; ARG-580; VAL-586; TYR-619; ALA-757 AND GLY-846.
REFERENCE 142 (residues 1 to 919)
AUTHORS Ahmed,S.F., Cheng,A., Dovey,L., Hawkins,J.R., Martin,H., Rowland,J., Shimura,N., Tait,A.D. and Hughes,I.A.
TITLE Phenotypic features, androgen receptor binding, and mutational analysis in 278 clinical cases reported as androgen insensitivity

syndrome
JOURNAL J. Clin. Endocrinol. Metab. 85 (2), 658-665 (2000)
MEDLINE 20152731
PUBMED 10690872
REMARK VARIANTS CAIS AND PAIS.
REFERENCE 143 (residues 1 to 919)
AUTHORS Chavez,B., Mendez,J.P., Ulloa-Aguirre,A., Larrea,F. and Vilchis,F.
TITLE Eight novel mutations of the androgen receptor gene in patients
with androgen insensitivity syndrome
JOURNAL J. Hum. Genet. 46 (10), 560-565 (2001)
MEDLINE 21470699
PUBMED 11587068
REMARK VARIANTS PAIS THR-682 AND GLU-711, VARIANTS CAIS GLU-743; VAL-827
AND ARG-874, AND VARIANT MAIS TYR-879.
REFERENCE 144 (residues 1 to 919)
AUTHORS Sills,E.S., Sholes,T.E., Perloe,M., Kaplan,C.R., Davis,J.G. and
Tucker,M.J.
TITLE Characterization of a novel receptor mutation A-->T at exon 4 in
complete androgen insensitivity syndrome and a carrier sibling via
bidirectional polymorphism sequence analysis
JOURNAL Int. J. Mol. Med. 9 (1), 45-48 (2002)
MEDLINE 21610990
PUBMED 11744994
REMARK VARIANT CAIS TYR-705.
COMMENT

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collaboration between the Swiss Institute of Bioinformatics and
the EMBL outstation - the European Bioinformatics Institute.
The original entry is available from <http://www.expasy.ch/sprot>
and <http://www.ebi.ac.uk/sprot>

[FUNCTION] THE STEROID HORMONES AND THEIR RECEPTORS ARE INVOLVED IN
THE REGULATION OF EUKARYOTIC GENE EXPRESSION AND AFFECT CELLULAR
PROLIFERATION AND DIFFERENTIATION IN TARGET TISSUES.

[SUBUNIT] Binds DNA as a homodimer. The AR N-terminal poly-Gln
region binds RAN resulting in enhancement of AR-mediated
transactivation. RAN binding decreases as the poly-Gln length
increases.

[SUBCELLULAR LOCATION] Nuclear.

[DOMAIN] Composed of three domains: a modulating N-terminal domain,
a DNA-binding domain and a C-terminal steroid-binding domain.

[POLYMORPHISM] The poly-Gln region of AR is highly polymorphic and
the number of Gln varies in the population (from 17 to 26). A
smaller size of the poly-Gln region may be associated with the
development of prostate cancer.

[POLYMORPHISM] The poly-Gly region of AR is also polymorphic and
ranges from 24 to 31 Gly.

[DISEASE] DEFECTS IN AR ARE THE CAUSE OF ANDROGEN INSENSIBILITY
SYNDROME (AIS), PREVIOUSLY KNOWN AS TESTICULAR FEMINIZATION
SYNDROME (TFM). IT CAN BE COMPLETE (CAIS) WHEN EXTERNAL GENITALIA
ARE PHENOTYPICALLY FEMALE; OR PARTIAL (PAIS) WHEN EXTERNAL
GENITALIA ARE SUBSTANTIVELY AMBIGUOUS OR MILD (MAIS) WHEN EXTERNAL
GENITALIA ARE NORMAL MALE OR NEARLY SO.

[DISEASE] DEFECTS IN AR ARE THE CAUSE OF X-LINKED SPINAL AND BULBAR
MUSCULAR ATROPHY (SBMA) (ALSO KNOWN AS KENNEDY'S DISEASE). IN SBMA
PATIENTS THE NUMBER OF GLN RANGES FROM 40 TO 52. LONGER EXPANSIONS
RESULT IN EARLIER ONSET AND MORE SEVERE CLINICAL MANIFESTATIONS OF
THE DISEASE.

[DISEASE] DEFECTS IN AR MAY PLAY A ROLE IN METASTATIC PROSTATE
CANCER. THE MUTATED RECEPTOR STIMULATES PROSTATE GROWTH AND

METASTASES DEVELOPMENT DESPITE OF ANDROGEN ABLATION. THIS TREATMENT CAN REDUCE PRIMARY AND METASTATIC LESIONS PROBABLY BY INDUCING APOPTOSIS OF TUMOR CELLS WHEN THEY EXPRESS THE WILD-TYPE RECEPTOR. [DISEASE] DEFECTS IN AR MAY BE THE CAUSE OF INFERTILITY MALE SYNDROME. IT IS CHARACTERIZED BY AZOOSPERMIA, ELEVATED TESTOSTERONE AND LUTEINIZING HORMONE PLASMA LEVELS AND AN ABNORMAL ANDROGEN RECEPTOR.

[MISCELLANEOUS] In the absence of ligand, steroid hormone receptors are thought to be weakly associated with nuclear components; hormone binding greatly increases receptor affinity. The hormone-receptor complex appears to recognize discrete DNA sequences upstream of transcriptional start sites.

[SIMILARITY] Belongs to the nuclear hormone receptor family. NR3 subfamily.

[DATABASE] NAME=Androgen receptor gene mutations database;

WWW='http://www.mcgill.ca/androgendb/';

FTP='ftp://ftp.ebi.ac.uk/pub/databases/androgen'.

FEATURES	Location/Qualifiers
source	1..919 /organism="Homo sapiens" /mol_type="unassigned DNA" /db_xref="taxon:9606"
gene	1..919 /gene="AR" /note="synonyms: NR3C4, DHTR"
<u>Protein</u>	1..919 /gene="AR" /product="Androgen receptor"
<u>Region</u>	1..558 /gene="AR" /region_name="Domain" /note="MODULATING."
<u>Region</u>	2 /gene="AR" /region_name="Variant" /note="E -> K (IN PAIS). /FTId=VAR_004679."
<u>Region</u>	54..57 /gene="AR" /region_name="Domain" /note="POLY-LEU."
<u>Region</u>	54 /gene="AR" /region_name="Variant" /note="L -> S (IN PROSTATE CANCER). /FTId=VAR_004680."
<u>Region</u>	57 /gene="AR" /region_name="Variant" /note="L -> Q (IN PROSTATE CANCER). /FTId=VAR_004681."
<u>Region</u>	58..89 /gene="AR" /region_name="Domain" /note="GLN-RICH."
<u>Region</u>	58..78 /gene="AR" /region_name="Domain" /note="POLY-GLN."
<u>Region</u>	64 /gene="AR" /region_name="Variant" /note="Q -> R (IN PROSTATE CANCER). /FTId=VAR_009711."

Region 74..78
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/note="MISSING. /FTId=VAR_004682."

Region 84..89
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/region_name="Domain"
/note="POLY-GLN."

Region 112
/gene="AR"
/region_name="Variant"
/note="Q -> H (IN PROSTATE CANCER). /FTId=VAR_009712."

Region 166
/gene="AR"
/region_name="Conflict"
/note="G -> A (IN REF. 3)."

Region 180
/gene="AR"
/region_name="Variant"
/note="K -> R (IN PROSTATE CANCER). /FTId=VAR_009713."

Region 193..197
/gene="AR"
/region_name="Domain"
/note="POLY-GLN."

Region 194
/gene="AR"
/region_name="Variant"
/note="Q -> R (IN CAIS). /FTId=VAR_009224."

Region 205
/gene="AR"
/region_name="Variant"
/note="S -> R. /FTId=VAR_009714."

Region 212
/gene="AR"
/region_name="Conflict"
/note="A -> R (IN REF. 5 AND 6)."

Region 214
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/region_name="Variant"
/note="G -> R (IN MAIS; 20% LOWER TRANSACTIVATION CAPACITY). /FTId=VAR_009715."

Region 255
/gene="AR"
/region_name="Variant"
/note="L -> P (IN CAIS). /FTId=VAR_009225."

Region 266
/gene="AR"
/region_name="Variant"
/note="M -> T (IN PROSTATE CANCER). /FTId=VAR_009716."

Region 269
/gene="AR"
/region_name="Variant"
/note="P -> S (IN PROSTATE CANCER). /FTId=VAR_009717."

Region 340
/gene="AR"
/region_name="Variant"
/note="P -> L (IN PROSTATE CANCER). /FTId=VAR_009718."

Region 372..381
/gene="AR"
/region_name="Domain"

Region /note="POLY-PRO."
390
/gene="AR"
/region_name="Variant"
/note="P -> R (IN CAIS). /FTId=VAR_009226."
Region 390
/gene="AR"
/region_name="Variant"
/note="P -> S (IN MAIS). /FTId=VAR_009227."
Region 396..402
/gene="AR"
/region_name="Domain"
/note="POLY-ALA."
Region 443
/gene="AR"
/region_name="Variant"
/note="Q -> R (IN CAIS; MIGHT BE A POLYMORPHISM).
/FTId=VAR_009228."
Region 449..472
/gene="AR"
/region_name="Domain"
/note="POLY-GLY."
Region 465..472
/gene="AR"
/region_name="Variant"
/note="MISSING. /FTId=VAR_004683."
Region 475
/gene="AR"
/region_name="Conflict"
/note="G -> E (IN REF. 4 AND 7)."
Region 491
/gene="AR"
/region_name="Variant"
/note="G -> S (IN CAIS). /FTId=VAR_009719."
Region 528
/gene="AR"
/region_name="Variant"
/note="D -> G (IN PROSTATE CANCER). /FTId=VAR_009720."
Region 547
/gene="AR"
/region_name="Variant"
/note="L -> F (IN PAIS). /FTId=VAR_009721."
Region 548
/gene="AR"
/region_name="Variant"
/note="P -> S (IN MAIS). /FTId=VAR_009722."
Site 559..624
/gene="AR"
/site_type="DNA binding"
/note="NUCLEAR RECEPTOR-TYPE."
Region 559..579
/gene="AR"
/region_name="Zinc finger region"
/note="C4-TYPE."
Region 559
/gene="AR"
/region_name="Variant"
/note="C -> Y (IN CAIS). /FTId=VAR_009723."
Region 565
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Region /region_name="Conflict"
/note="E -> K (IN REF. 8)."
568
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/region_name="Variant"
/note="G -> V (IN PAIS). /FTId=VAR_009725."
Region 568
/gene="AR"
/region_name="Variant"
/note="G -> W (IN PAIS). /FTId=VAR_009726."
Region 571
/gene="AR"
/region_name="Variant"
/note="Y -> C (IN CAIS). /FTId=VAR_009727."
Region 573
/gene="AR"
/region_name="Variant"
/note="A -> D (IN CAIS; DEFECTIVE DNA BINDING AND
TRANSACTIVATION). /FTId=VAR_009728."
Region 574
/gene="AR"
/region_name="Variant"
/note="L -> P (IN PROSTATE CANCER). /FTId=VAR_009729."
Region 575
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/note="T -> A (IN PROSTATE CANCER). /FTId=VAR_009730."
Region 576
/gene="AR"
/region_name="Variant"
/note="C -> F (IN CAIS; LACK OF DNA BINDING).
/FTId=VAR_009731."
Region 576
/gene="AR"
/region_name="Variant"
/note="C -> R (IN CAIS). /FTId=VAR_009732."
Region 579
/gene="AR"
/region_name="Variant"
/note="C -> F (IN CAIS; REDUCED TRANSCRIPTION AND DNA
BINDING). /FTId=VAR_009733."
Region 579
/gene="AR"
/region_name="Variant"
/note="C -> Y (IN CAIS). /FTId=VAR_009734."
Region 580
/gene="AR"
/region_name="Variant"
/note="K -> R (IN PROSTATE CANCER). /FTId=VAR_009735."
Region 581
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Region 582
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Region 582
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Region 585
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/note="R -> K (IN CAIS). /FTId=VAR_009740."
Region 586
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/note="A -> V (IN PROSTATE CANCER; SOMATIC MUTATION).
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Region 587
/gene="AR"
/region_name="Variant"
/note="A -> S (IN PROSTATE CANCER; SOMATIC MUTATION).
/FTId=VAR_009742."
Region 595..619
/gene="AR"
/region_name="Zinc finger region"
/note="C4-TYPE."
Region 596
/gene="AR"
/region_name="Variant"
/note="A -> T (IN PAIS; ABOLISHES DIMERIZATION).
/FTId=VAR_009743."
Region 597
/gene="AR"
/region_name="Variant"
/note="S -> G (IN PAIS; HIGH DISSOCIATION RATE; ASSOCIATED
WITH P-617 IN A PAIS PATIENT; PARTIALLY RESTORES
DNA-BINDING ACTIVITY OF P-617 MUTANT RECEPTORS).
/FTId=VAR_009744."
Region 597
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/region_name="Variant"
/note="S -> T (IN PAIS). /FTId=VAR_009745."
Region 601
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Region 607
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/FTId=VAR_004684."
Region 608
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NUCLEAR LOCALIZATION). /FTId=VAR_004685."
Region 610
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611
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Region 615
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Region 615
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Region 615
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Region 616
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Region 617
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ACTIVITY; ASSOCIATED WITH G-597 IN A PAIS PATIENT).
/FTId=VAR_009755."
Region 619
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Region 629
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Region 630
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Region 645
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Region 647
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Region 664
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Region 670
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Region 683
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Region 684
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Region 686
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Region 690..919
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Region 692
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Region 705
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Region 710
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Region 717
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Region 720
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METASTASES). /FTId=VAR_009783."
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754
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Region 757
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Region 758
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Region /FTId=VAR_009809."
759
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Region 759
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Region /note="S -> P (IN PROSTATE CANCER). /FTid=VAR_009811."
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Region 765
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Region 772
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779
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780
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782
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784
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787
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788
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790
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791
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793
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794
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798
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806
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807
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807
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Region 812
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Region 814
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Region 820
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Region 821
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Region 827
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Region 831
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Region 840
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Region /note="R -> H (IN PAIS). /FTId=VAR_004723."
840
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Region 842
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Region 854
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Region 855
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Region 855
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Region 856
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Region 863
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Region 864
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Region 864
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Region 865
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Region 866
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Region 866
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869
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Region 871
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Region 874
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Region 874
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Region 877
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CHARACTERISTICS AND RESPONSE TO ANTIANDROGENS; FOUND IN
BONE METASTASES). /FTId=VAR_004732."
Region 877
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Region 879
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Region 880
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Region 881
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Region 886
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Region 889
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Region 890
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Region 891
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Region 892
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Region 895
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Region 896
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Region 898
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Region 902
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Region 903
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Region 904
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Region 904
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Region 910
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/region_name="Variant"

Region 911
/note="K -> R (IN PROSTATE CANCER). /FTId=VAR_009859."
/gene="AR"
/region_name="Variant"

Region 913
/note="V -> L (IN PAIS). /FTId=VAR_009860."
/gene="AR"

//

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